hepatic lipase deficiency

Hepatic lipase deficiency is a disorder that affects the body's ability to break down fats (lipids). People with this disorder have increased amounts of certain fats, known as triglycerides and cholesterol, in the blood. These individuals also have increased amounts of molecules known as high-density lipoproteins (HDLs) and decreased amounts of molecules called low-density lipoproteins (LDL). These molecules transport triglycerides and cholesterol throughout the body. In people with hepatic lipase deficiency, the LDL molecules are often abnormally large.

Normally, high levels of HDL (known as "good cholesterol") and low levels of LDL (known as "bad cholesterol") are protective against an accumulation of fatty deposits on the artery walls (atherosclerosis) and heart disease. However, some individuals with hepatic lipase deficiency, who have this imbalance of HDL and LDL, develop atherosclerosis and heart disease in mid-adulthood, while others do not. It is unknown whether people with hepatic lipase deficiency have a greater risk of developing atherosclerosis or heart disease than individuals in the general population. Similarly, it is unclear how increased blood triglycerides and cholesterol levels affect the risk of atherosclerosis and heart disease in people with hepatic lipase deficiency.

Frequency

Hepatic lipase deficiency is likely a rare disorder; only a few affected families have been reported in the scientific literature.

Genetic Changes

Hepatic lipase deficiency is caused by mutations in the *LIPC* gene. This gene provides instructions for making an enzyme called hepatic lipase. This enzyme is produced by liver cells and released into the bloodstream where it helps convert very low-density lipoproteins (VLDLs) and intermediate-density lipoproteins (IDLs) to LDLs. The enzyme also assists in transporting HDLs that carry cholesterol and triglycerides from the blood to the liver, where the HDLs deposit these fats so they can be redistributed to other tissues or removed from the body.

LIPC gene mutations prevent the release of hepatic lipase from the liver or decrease the enzyme's activity in the bloodstream. As a result, VLDLs and IDLs are not efficiently converted into LDLs, and HDLs carrying cholesterol and triglycerides remain in the bloodstream. It is unclear what effect this change in lipid levels has on people with hepatic lipase deficiency.

Inheritance Pattern

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Other Names for This Condition

- HL deficiency
- hyperlipidemia due to hepatic triglyceride lipase deficiency
- LIPC deficiency

Diagnosis & Management

Genetic Testing

- Genetic Testing Registry: Hepatic lipase deficiency
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C3151466/

Other Diagnosis and Management Resources

- MedlinePlus Encyclopedia: Cholesterol Testing and Results
  https://medlineplus.gov/ency/patientinstructions/000386.htm
- MedlinePlus Encyclopedia: Triglyceride Level
  https://medlineplus.gov/ency/article/003493.htm

General Information from MedlinePlus

- Diagnostic Tests
  https://medlineplus.gov/diagnostictests.html
- Drug Therapy
  https://medlineplus.gov/drugtherapy.html
- Genetic Counseling
  https://medlineplus.gov/geneticcounseling.html
- Palliative Care
  https://medlineplus.gov/palliativecare.html
- Surgery and Rehabilitation
  https://medlineplus.gov/surgeryandrehabilitation.html

Additional Information & Resources

MedlinePlus

- Encyclopedia: Cholesterol Testing and Results
  https://medlineplus.gov/ency/patientinstructions/000386.htm
- Encyclopedia: Triglyceride Level
  https://medlineplus.gov/ency/article/003493.htm
- Health Topic: Cholesterol
  https://medlineplus.gov/cholesterol.html
- Health Topic: Triglycerides
  https://medlineplus.gov/triglycerides.html

Genetic and Rare Diseases Information Center

- Hepatic lipase deficiency
  https://rarediseases.info.nih.gov/diseases/12864/hepatic-lipase-deficiency

Additional NIH Resources

- National Heart, Lung, and Blood Institute: What is Cholesterol?
  https://www.nhlbi.nih.gov/health/health-topics/topics/hbc

Educational Resources

- American Heart Association: Good vs. Bad Cholesterol
  http://www.heart.org/HEARTORG/Conditions/Cholesterol/AboutCholesterol/Good-vs-Bad-Cholesterol_UCM_305561_Article.jsp
- Centers for Disease Control and Prevention: LDL and HDL: "Bad" and "Good" Cholesterol
  https://www.cdc.gov/cholesterol/ldl_hdl.htm
- Cleveland Clinic: Cholesterol Facts and Fiction
  http://my.clevelandclinic.org/health/articles/cholesterol-facts-and-fiction
- CLIMB: Hepatic Lipase Deficiency Info Sheet
  http://www.climb.org.uk/IMD/Hotel/HepaticLipaseDeficiency.pdf
- Disease InfoSearch: Hepatic lipase deficiency
  http://www.diseaseinfosearch.org/Hepatic+lipase+deficiency/8508
- Lucile Packard Children's Hospital: Cholesterol, LDL, HDL, and Triglycerides in Children and Adolescents
  http://www.stanfordchildrens.org/en/topic/default?id=cholesterol-ldl-hdl-and-triglycerides-in-children-and-adolescents-90-P01593
- MalaCards: hepatic lipase deficiency
  http://www.malacards.org/card/hepatic_lipase_deficiency

- Merck Manual Professional Version: Dyslipidemia
  http://www.merckmanuals.com/professional/endocrine-and-metabolic-disorders/lipid-disorders/dyslipidemia
- Orphanet: Hyperlipidemia due to hepatic triglyceride lipase deficiency
  http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=140905

Patient Support and Advocacy Resources

- American Heart Association
  http://www.heart.org/HEARTORG/
- CLIMB: Children Living with Inherited Metabolic Diseases (UK)
  http://www.climb.org.uk/
- National Lipid Association
  https://www.lipid.org/
- World Heart Federation
  http://www.world-heart-federation.org/

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28hepatic+lipase+deficiency%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D

OMIM

- HEPATIC LIPASE DEFICIENCY
  http://omim.org/entry/614025

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